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1: AAA79207. inositol polyphos...[gi:1019103]

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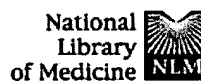
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 DEFINITION inositol polyphosphate 5-phosphatase.
 ACCESSION AAA79207
 VERSION AAA79207.1 GI:1019103
 DBSOURCE locus HUMINP5P accession M74161.1
 KEYWORDS .
 SOURCE Homo sapiens (human)
 ORGANISM Homo sapiens
 Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
 Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.
 REFERENCE 1 (residues 1 to 942)
 AUTHORS Jefferson, A.B. and Majerus, P.W.
 TITLE Properties of type II inositol polyphosphate 5-phosphatase
 JOURNAL J. Biol. Chem. 270 (16), 9370-9377 (1995)
 MEDLINE 95238452
 PUBMED 7721860
 COMMENT Method: conceptual translation.
 FEATURES Location/Qualifiers
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 901 enilasifgs lllrnpaghq kldmtekkka gefihqflcn pl

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☐ 1: Hum Mol Genet 1995 Dec;4(12):2245-50

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Lowe syndrome, a deficiency of phosphatidylinositol 4,5-bisphosphate 5-phosphatase in the Golgi apparatus.

Suchy SF, Olivos-Glander IM, Nussabaum RL.

PubMed
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Laboratory of Genetic Disease Research, National Center for Human Genome Research, National Institutes of Health, Bethesda, Maryland 20892, USA.

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The oculocerebrorenal syndrome of Lowe (OCRL) is an X-linked disorder characterized by congenital cataracts, renal tubular dysfunction and neurological deficits. The gene responsible for this disorder, OCRL-1, has been cloned and mutations identified in patients. The gene product (ocrl-1) has extensive sequence homology to a 75 kDa inositol polyphosphate 5-phosphatase. We report here that OCRL patients' fibroblasts show no abnormality in inositol polyphosphate 5-phosphatase activity, but are deficient in a phosphatidylinositol 4,5-bisphosphate [PtdIns(4,5)P₂] 5-phosphatase activity localized to the Golgi apparatus. Direct biochemical diagnosis of this human disease should now be possible. PtdIns(4,5)P₂ has been implicated in Golgi vesicular transport through its role in the regulation of ADP-ribosylation factor, phospholipase D and actin assembly in the cytoskeleton. The regulation of PtdIns(4,5)P₂ levels by PtdIns(4,5)P₂ 5-phosphatase may, therefore, be important in the modulation of Golgi vesicular transport. Given that the primary defect in OCRL is a deficiency of a Golgi PtdIns(4,5)P₂ phosphatase, we hypothesize that the disorder results from dysregulation of Golgi function and in this way causes developmental defects in the lens and abnormal renal and neurological function.

PMID: 8634694 [PubMed - indexed for MEDLINE]

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☐ 1: AAB03216. phosphatidylinosi...[gi:1399105]

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LOCUS AAB03216 397 aa linear PRI 29-JUN-1996
 DEFINITION phosphatidylinositol (4,5)bisphosphate 5-phosphatase homolog.
 ACCESSION AAB03216
 VERSION AAB03216.1 GI:1399105
 DBSOURCE locus HSU45975 accession [U45975.1](#)
 KEYWORDS .
 SOURCE Homo sapiens (human)
 ORGANISM Homo sapiens
 Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
 Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.
 REFERENCE 1 (residues 1 to 397)
 AUTHORS Nussbaum, R.L.
 TITLE Direct Submission
 JOURNAL Submitted (11-JAN-1996) Robert L. Nussbaum, NCHGR, NIH, 49 Convent
 Drive, Bethesda, MD 20892, USA
 COMMENT Method: conceptual translation.
 FEATURES Location/Qualifiers
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 /db_xref="taxon:9606"
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 and R15390"
 Protein 1..397
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 5-phosphatase homolog"
 CDS 1..397
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Build 2.2.2002.1.10.1



Sequence Revision History

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Sequence Revision History				
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1420920	13254464	2	<u>Mar 8 2001 17:15</u>	Dead
1420920	1420920	1	<u>Jul 16 1996 0:12</u>	Dead

SeqId gi|1420920 was first seen at NCBI on Jul 16 1996 0:12

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☐ 1: [AAB03839](#)

gb|AAB03839.1|[1420920]

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